Assessing genetic risk: learning from mistakes made in screening for sickle cell disorders in England

ESCR Seminar 2: Using Assessments of Biological and Genetic Risk to Inform Policy Priorities. 1st June 2005, University of Leeds.

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A reflection on some historical influences influencing my own perceptions on ethnicity and screening for sickle cell disorders

Anionwu EN. Sickle Cell & Thalassaemia. Community experiences and official response. In: Ahmad WIU (ed.) *'Race' and Health in Contemporary Britain.* Open University Press, 1993: 76-95.

Anionwu EN. Ethnic Origin of Sickle & Thalassaemia Counsellors: Does it Matter? In Kelleher D & Hillier S (eds) *Researching Cultural Differences in Health*. Routledge, London,1996:160-189.

Atkin K, Ahmad W, Anionwu EN. Screening and counselling for sickle cell disorders and thalassaemia: the experience of parents and health professionals. *Social Science and Medicine.* 47 (11):1639-1651, 1998

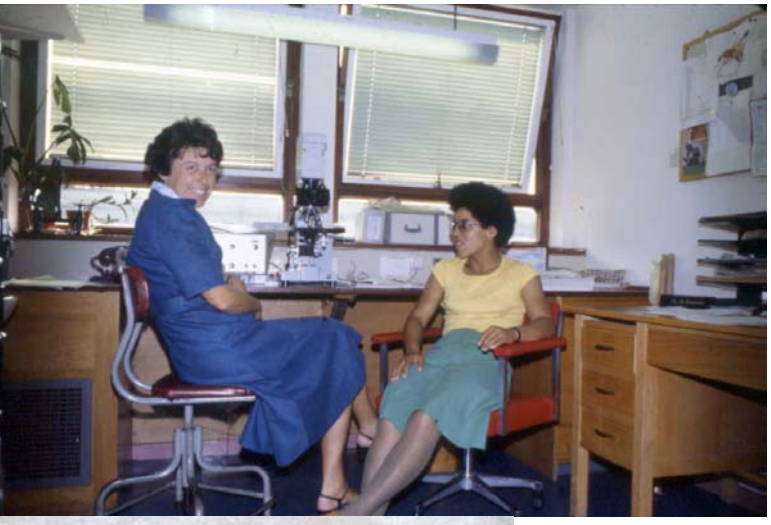
Anionwu E.N. and Atkin K. *The Politics of Sickle Cell and Thalassaemia*. Buckingham: Open University Press, June 2001

Anionwu E.N. (2003) Genetic screening for sickle cell and thalassaemia: can we learn anything from the UK experience? Chapter 11, pages 105-112 in *Key Issues In Bioethics. A guide for teachers.* Eds: R Levinson & M Reiss. London: RoutledgeFalmer.

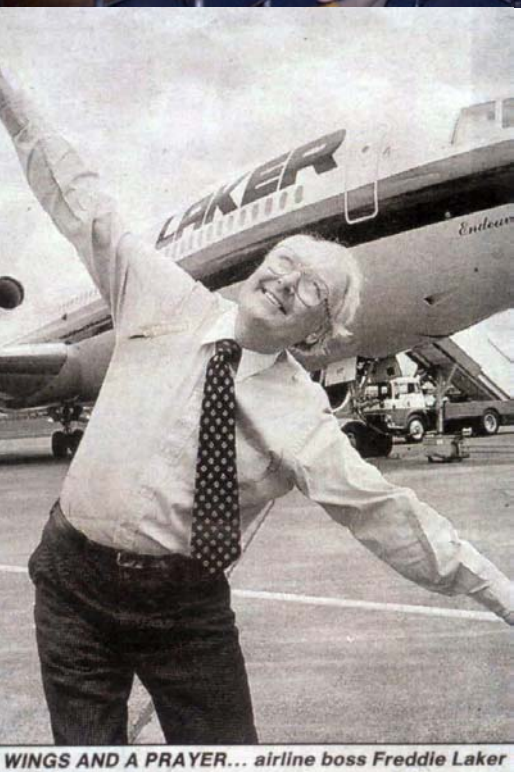
Aspinall, PJ., Dyson, SM., and Anionwu EN. (2003) The feasibility of using ethnicity as a primary tool for antenatal selective screening for sickle cell disorders: pointers from the research evidence. *Social Science and Medicine*; 56: 285-297

Anionwu, E.N. (2004) Sickle cell disorders and thalassaemia: the challenge for health professionals and resources available. Chapter 25, pages 227-234 in *Practical Management of Haemoglobinopathies*, Editor I. E. Okpala. Oxford: Blackwell Publishing Ltd.

Brent and the USA: the late 1970's



Dr Milica Brozovic & EA – set up 1st Sickle Cell/Thal Centre in Brent 1979



WINGS AND A PRAYER... airline boss Freddie Laker

EA on her 1st ever SCD/Thal training course in San Francisco

MARVELLE, ELIZABETH AND NINA COUNSELLORS FOR SICKLE CELL AND THALASSAEMIA

Photo/Power Archive Photocopy

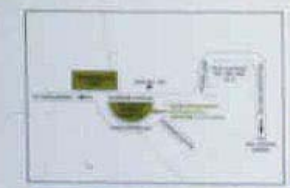
COUNSELLING AND ADVICE

BRENT SICKLE CELL CENTRE

INFORMATION &

SCREENING

BRENT SICKLE CELL CENTRE
WILLESDEN HOSPITAL
HARLESDEN ROAD
LONDON NW10 3RY



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A multi-ethnic approach

A report of the Royal College of Physicians, entitled *Prenatal Diagnosis and Genetic Screening: Community and service implications*, describes community genetic services as involving population screening, counselling before, during or after pregnancy, and the management of abortion or foetal abnormality.

It goes on to state: "Two to three per cent of couples are at high and recurrent risk of having children with an inherited disorder. It is increasingly possible to detect these carriers by biochemical or DNA methods, and whenever possible this should be done prior to a pregnancy being established so that the couple can choose from the full range of options which could be available. These developments draw the whole community within the range of

In the first of two articles, Elizabeth Anionwu outlines the characteristics of four genetic conditions recognised as important to the delivery of community genetic services to multi-ethnic communities in Britain.

ETHNIC ORIGIN OF MOTHERS WITH Hb AS (CMH: 1985)

Slide courtesy Joan Henthorn, Central Middlesex Hospital

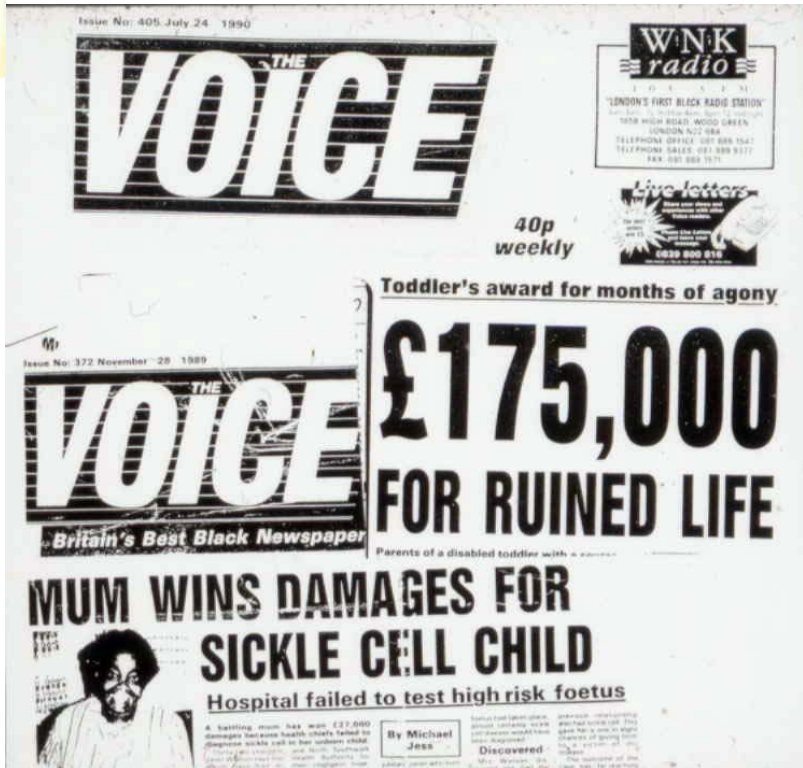
Screening Policy:	Selective	Universal
	Jan-June	July-Dec
Afro-Caribbean	22	27
Indian		4
Other		8
Total	22	39

Antenatal

"There must be a skeleton in my family." White caller to BBC London phone-in following item on work of Brent Sickle Support Group, 1978

Community development philosophy

Medico-legal cases supported by Sickle Cell Society



"Sickle-cell anaemia is not of great consequence to us in the context of genetic counselling in the United Kingdom. The sickling trait and sickle cell anaemia appear to be confined to peoples of African and Eastern origin".

Stevenson A.C. and Davidson B.C.C. (1976) *Genetic counselling*. London: William Heinemann, page 274.

Ethnicity as a screening tool?

1974

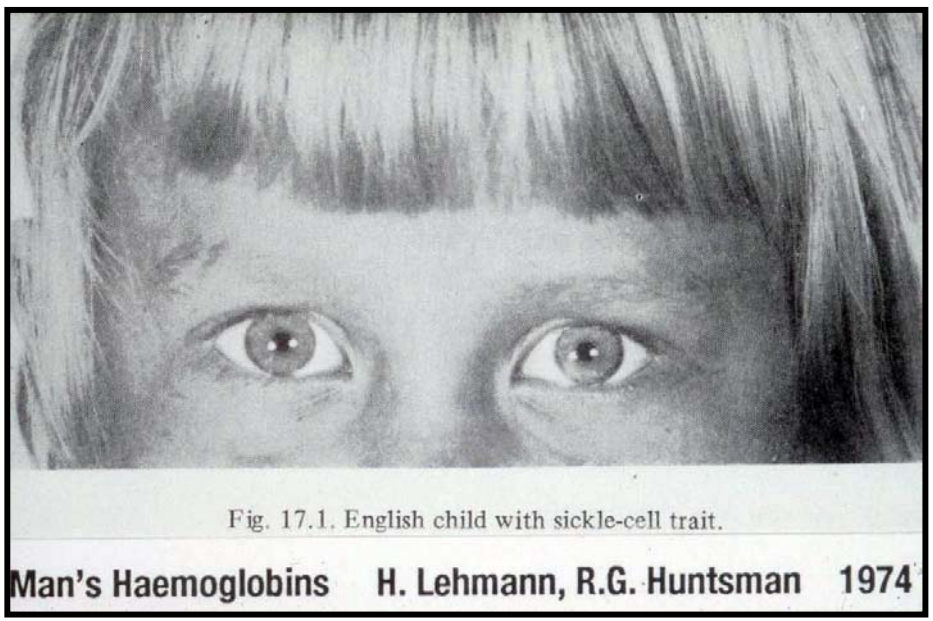


Fig. 17.1. English child with sickle-cell trait.

Man's Haemoglobins H. Lehmann, R.G. Huntsman 1974

1996

SUNDAY TELEGRAPH P14 JUNE 9 1996

Black disease gene found in white mothers

by VICTORIA MACDONALD

Health Correspondent

WHITE people are searching family trees after being told they are carrying the gene of disease which was believed to have afflicted only black communities.

The introduction of a screening programme in some parts of the country has disclosed a handful of Caucasians with the genetic trait of sickle cell anaemia — an abnormality of the red blood cells.

Campaigners are now calling for nationwide universal screening in which all mothers, rather than those assumed to be at risk, are tested.

In some cases the disease can be fatal. A national sickle cell awareness day on

Unlikely carrier: Blonde, blue-eyed mother-to-be Michelle Milne has been found to have the sickle cell gene

Difficulties in using ethnicity as a screening criteria in SCD: north of England study[1]

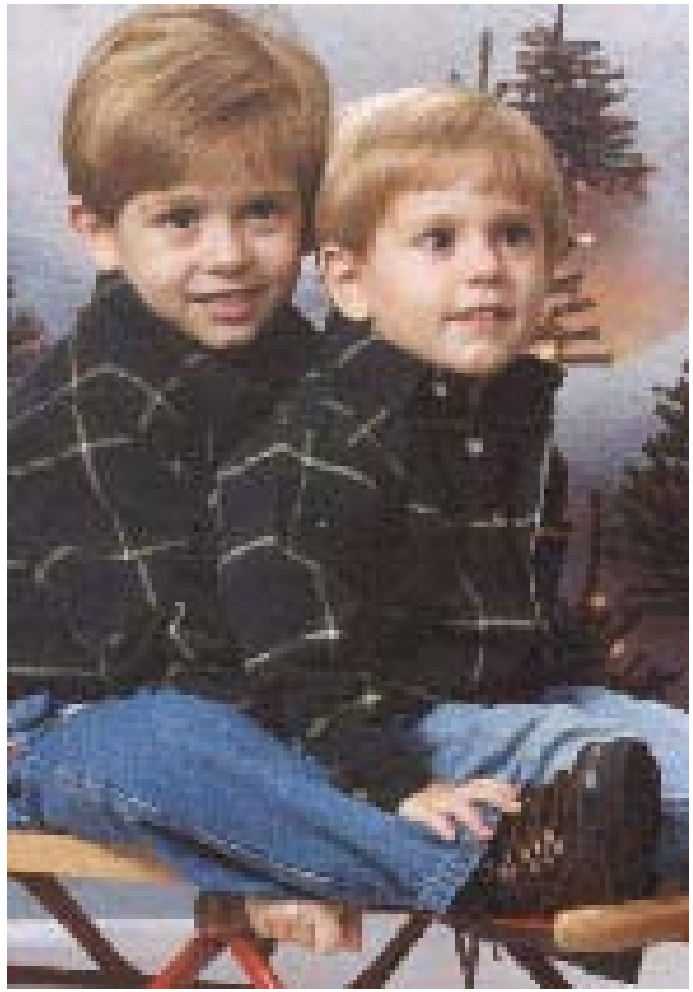
- problems of practitioners in determining ethnicity: 'They kept telling me, all superior like, "Oh no, you are the wrong ethnic background".'
- Identifying people of 'Afro-Caribbean origin by their surname.'
- resultant delays in screening

[1]Atkin, Ahmad & Anionwu, 1998 *Soc. Sci. Med.* 47:11 pp. 1639-1651

Source: http://www.scinfo.org/ptstory.htm (2002)

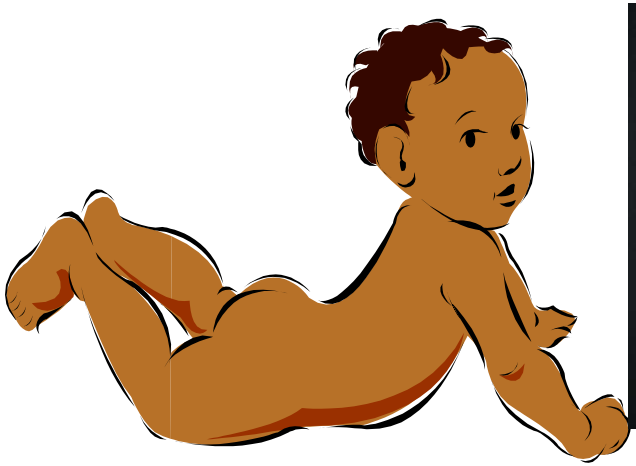
"Sickle Cell Disease can affect ANY race! My name is Vikki and my husband and I am a Caucasian couple with 2 beautiful boys that have Sickle Beta Thalassemia Plus. I am of Italian decent and have a thalassemia trait.

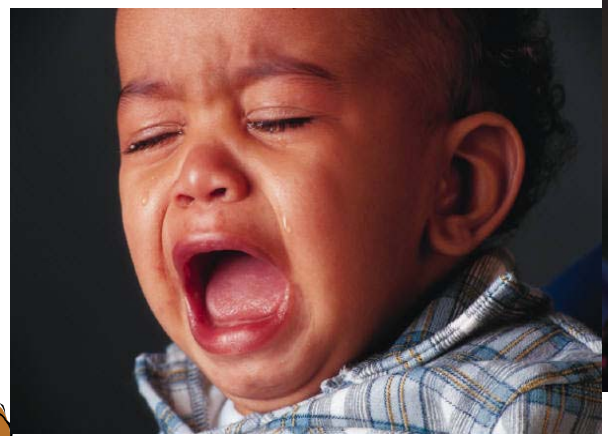
When I was pregnant I was tested and my doctor wanted to test my husband for it as well. I received a call several days later that he has a sickle trait. I said 'No way! My husband has blonde hair and blue eyes'. We *thought* at the time that this disease was ***only*** a black disease".



My two children both with Sickle Cell Disease type Sickle beta + thalassemia

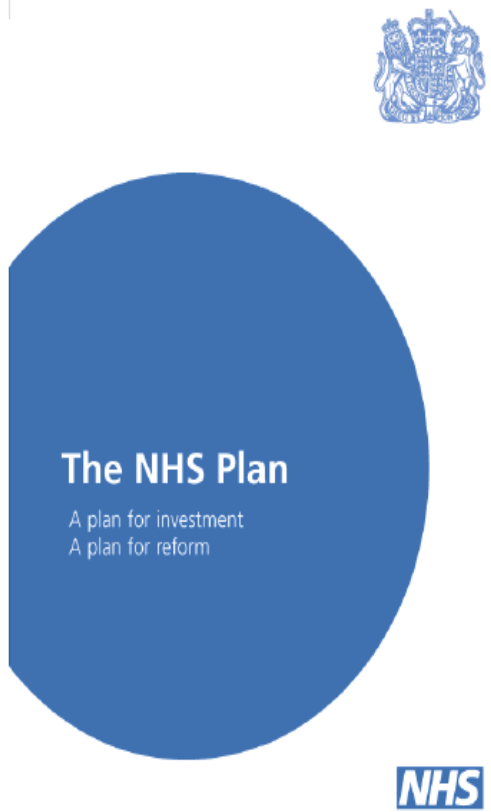
Heated debates in the UK on who should be screened for SCD





The NHS Plan - July 2000

- A linked **antenatal** and **neonatal** screening programme for Sickle Cell Disease and Thalassaemia was announced in the NHS Plan in September 2000, to be introduced by 2004. A development programme led by Dr Allison Streetly has started.



Aim of NHS newborn screening for Sickle Cell Disorders

- Aim: To achieve the lowest possible childhood mortality rates and to minimise morbidity from sickle cell disease in childhood.
- **Policy decision: All babies, regardless of ethnic origin, to be screened for SCD at time of the Guthrie heel prick test at 6-8 days. Informed by experience discussed at an international workshop.**

For updates on implementation visit www-phm.umds.ac.uk/haemscreening/

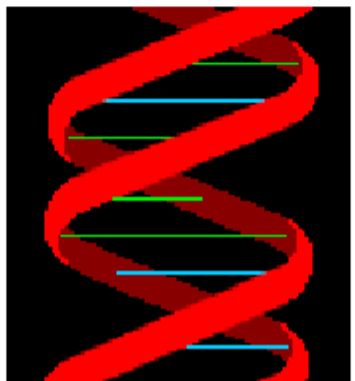
NHS Sickle Cell & Thalassaemia Screening Programme

Welcome to the NHS Sickle Cell & Thalassaemia Screening Programme Homepage.

'The NHS Sickle Cell & Thalassaemia Screening Programme aims to establish high quality newborn screening programmes for sickle cell disorders and antenatal screening programmes for sickle cell and thalassaemia. It is committed to ensuring that informed decision-making by women and their families is integral to the programme.'

Please use the following links to explore this site

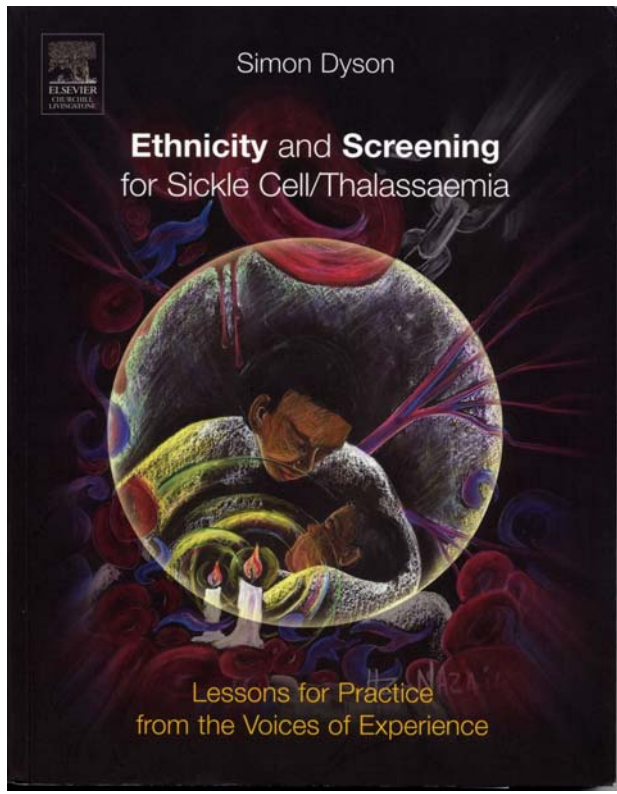
- Introduction
- Contact Us
- Steering Group
- Publications- including London Care Pathways report & RSCG Briefings
- Whats New? including Interim Awareness Training Workshops



- Implementation Plan
- Contact Us
- Links
- Workshops
- Job Vacancies & Recruitment

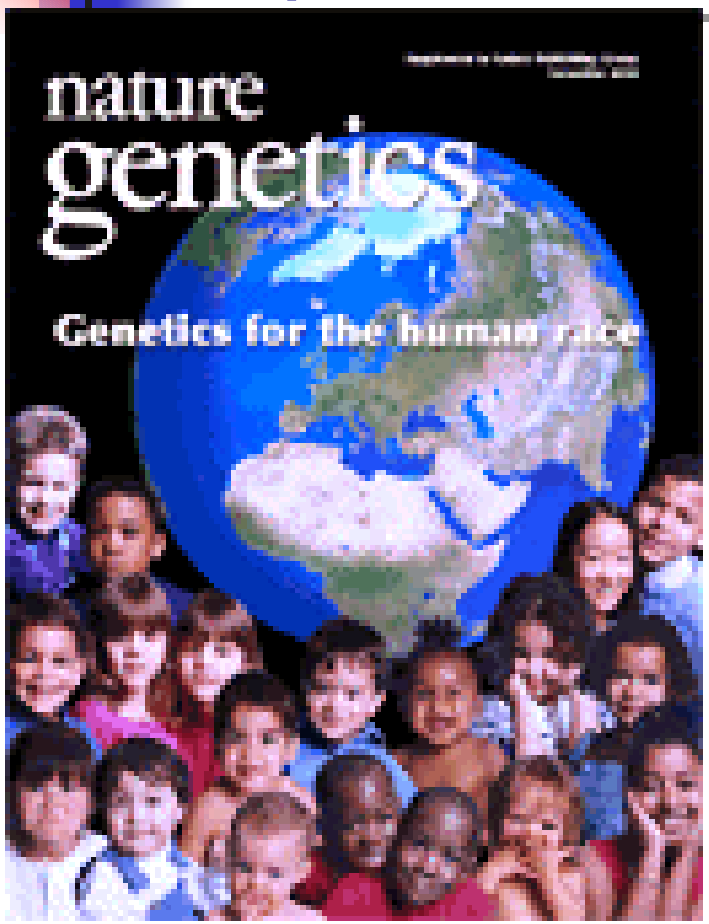
Implications for Practice – Reactions of majority ethnic groups*

- 'Usually, the reaction is, well this is usually a black blood problem, so how comes you're telling me, I'm white, that I am a carrier for this thing? And usually shock. And in some cases, it's anger, disbelief, denial'. Page113
- "Ooh I've got sickle." You know, it was like a party piece to tell her friends. It's like being cool, you know, 'I've probably got a bit of black somewhere'. Page 107.



*Dyson, S. (2005) *Ethnicity and Screening for Sickle Cell | Thalassaemia. Lessons for Practice from the Voices of Experience.* Edinburgh: Elsevier Churchill Livingstone

A final thought: could the SCD experience inform the wider debate?



Nature Genetics **36**, S54 - S60 (2004)
Published online: ; | doi:10.1038/ng1440
Implications of correlations between skin color and genetic ancestry for biomedical research
E J Parra[1], R A Kittles[2] & M D Shriver[3,]

November 2004, Volume 36 No 11s

Source: www.nature.com